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Superior Mesenteric Vein Thrombosis in a Young Female with Heterozygous Factor V Mutation: A Case Report

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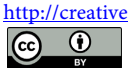

Abstract

Superior mesenteric vein thrombosis (SMVT) is a rare medical and surgical emergency. The majority of the patients present with non-specific signs and symptoms that include non-specific abdominal pain, nausea, vomiting, or hematochezia, which leads to the disease being overlooked even in acute settings. We present a case of SMVT in a young female heterozygous for factor V Leiden with associated risk factors that lead to a timely diagnosis with good outcomes.

Subject Areas

Pathology, Women's Health

Keywords

Thrombophilia, Factor V Leiden, Oral Contraceptive Pill (OCP), Smoking Tobacco, Abdominal Pain, Superior Mesenteric Vein Thrombosis

1. Introduction

The majority of cases of abdominal ischemia are arterial, and patients rarely present with venous thrombosis. Superior mesenteric vein (SMV) thrombosis is one of the rare etiology for intestinal ischemia and can be chronic, subacute, or acute. SMV thrombosis is usually secondary to thrombophilias acquired or inherited due

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to an underlying proinflammatory state [1]. The timely diagnosis in acute cases is critical; delay in seeking treatment can increase the chances of increased morbidity and mortality.

2. Case Presentation

A 33-year-old female presented to the emergency department (ED) with complaints of abdominal pain for the past 1 week. The patient reported that pain is 10/10, located in the epigastrium, intermittent, initially started when she was driving, progressive, radiating to her back, and worse on deep inspiration. Her past medical and surgical history is significant for depression and a cesarian section 7 years ago. Her last menstrual period (LMP) was 20 days prior with normal flow for 5 days.

Initially, the patient went to the urgent care center for evaluation. The patient was given a proton pump inhibitor on suspicion of gastroesophageal reflux disease, but her electrocardiogram (EKG) changes were suspicious, for which she was sent to the emergency department for detailed evaluation. She also endorsed decreased appetite with chronic constipation but denies any chest pain, shortness of breath, nausea, vomiting, diarrhea, or weight changes.

The patient is a smoker with 7.5 pack years and social alcohol intake. She has taken venlafaxine for a few years for her depression, and 6 months ago, she was started on ethinylestradiol-levonorgestrel by her primary care physician for contraception.

In the ED, her vitals showed a temperature of 97F, heart rate of 60 bpm, respiratory rate of 16/min, and blood pressure of 134/77 mmHg with SpO_2 of 99% on room air. Her physical examination showed a morbidly obese female with a BMI of 40.6 kg/m^2, moist mucous membrane, abdominal tenderness in all quadrants, more pronounced in the epigastric region, sluggish bowel sounds, guarding without rebound, lungs clear to auscultation bilaterally, bradycardia on cardiac auscultation with no murmurs, rubs or gallops.

Our differentials were acute coronary syndrome, myocardial infarction, pancreatitis, cholecystitis, hernia, splenic vein thrombosis, and superior mesenteric vein thrombosis.

The patient was given intravenous morphine along with intravenous hydration and was admitted for further workup. Her EKG showed sinus rhythm with a rate of 60, with a QTc of 412. The chest X-ray didn't show any acute pathology. Her SARS and influenza A/B antigens were negative. Her initial labs are given below (Table 1).

Table 1. Laboratory work-up.

Laboratory values	Patient value	Normal Value
Sodium	136 mmol/L	136 - 145 mmol/L
Potassium	3.6 mmol/L	3.5 - 5.3 mmol/L
White Blood Cells (WBCs)	12.10 × 3/uL	4.5 - 11 10 × 3/uL

Continued

Creatinine	0.9 mg/dl	0.5 - 1.0 mg/dl
Alanine Transaminase	28 U/L	6 - 29 U/L
Aspartate transaminase	14 U/L	10 - 36 U/L
Lipase	39 U/L	73 - 393 U/L

At this time, acute coronary syndrome and pancreatitis were ruled out. CT of the abdomen pelvis with intravenous contrast was done, which showed thrombosis of the superior mesenteric vein with associated thickening of the surrounding mesenteric fat extending down to the level of the right lower quadrant with a small amount of retroperitoneal fat. Multiple left ovarian cysts. Images are given below (Figure 1, Figure 2).

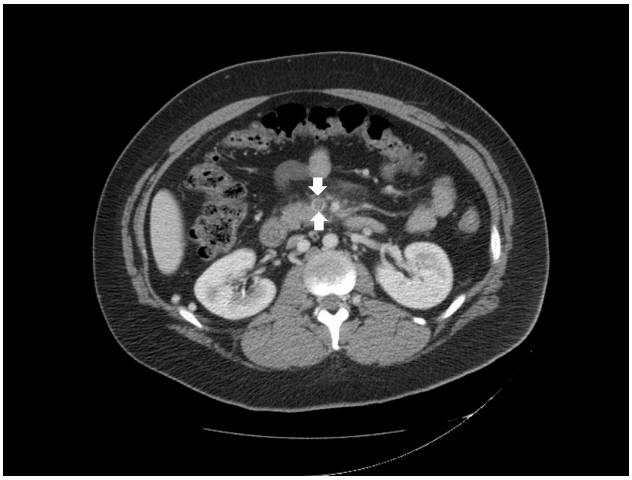

Figure 1. Thrombophlebitis of the superior mesenteric vein and its branches with associated mesenteric fat thickening.

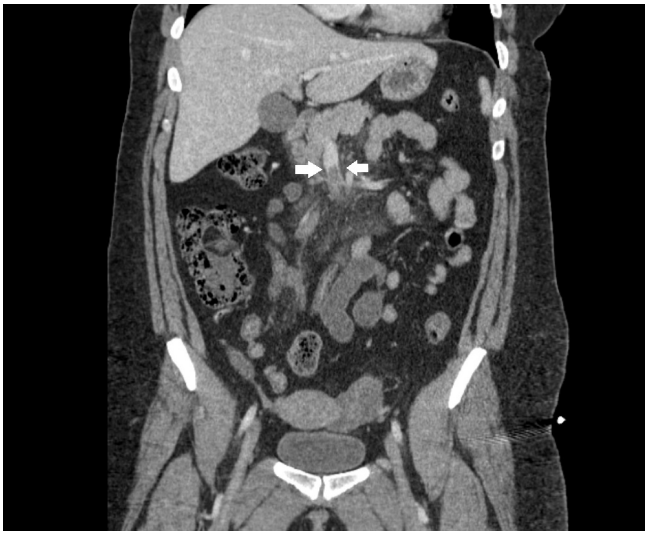

Figure 2. Thrombophlebitis of the superior mesenteric vein and its branches with associated mesenteric fat thickening.

The diagnosis was confirmed at this point with the help of the imaging. The patient was started on therapeutic anticoagulation with heparin. Surgery was consulted, and it was recommended to keep the patient nil per oral and to recommend outpatient evaluation from hematology. The patient stayed inpatient for 5 days, and her pain settled. Ultimately, a diet was introduced, which she tolerated well. The patient was discharged on anti-coagulation with apixaban and was referred to hematology with advice to quit smoking by discontinuing the ethinylestradiol-levonorgestrel pills and switching to an alternative form of contraception after consulting gynecology.

The hypercoagulability workup in the hematology office, including anticardiolipin antibodies IgA, IgG, IgM, beta-2 glycoprotein antibodies, lupus anticoagulant, factor II DNA analysis, protein C and S, and antithrombin panel was found to be negative. The patient was positive for c.1601G>A heterozygous variant factor V Leiden, and she was asked to continue anticoagulation for life. A repeat CT scan after 6 months of the abdomen pelvis with contrast showed the resolution of thrombosis.

3. Discussion

Superior mesenteric vein thrombosis (SMVT) is a rare yet potentially catastrophic cause of acute mesenteric ischemia. It carries a high risk of extensive intestinal infarction and an overall 30-day mortality rate of 20% - 32% [1]. The incidence of SMVT is reported to be 1.8 per 100,000 person-years which makes it a rare etiology. Patients typically present within the age range of 45 - 60 years, with a slight male predominance.

Mesenteric vein thrombosis (MVT) generally accounts for 1 in 5000 – 15,000 of all inpatient admissions, 1 in 1000 emergency department admissions, and 5% to 15% of all intestinal ischemic events, mainly involving the superior mesenteric vein [2] [3].

Risk factors for SMVT can be categorized according to Virchow's triad of direct Injury, stasis, and hypercoagulability. Direct Injury factors involve intra-abdominal inflammatory conditions such as pancreatitis, appendicitis, and diverticulitis, as well as previous intra-abdominal surgeries. Stasis-related risk factors include portal hypertension and obesity, which impair blood flow within the mesenteric venous system. Hypercoagulability is one of the most prevalent risk factors for MVT, which includes both acquired and inherited thrombophilia [2]. Studies indicate that specific hypercoagulable states are present in 60% - 75% of patients with MVT [4]. Furthermore, the prevalence of inherited thrombophilia among MVT patients has been estimated to range from 36% - 55% [5]. A study comparing patients with MVT to healthy individuals identified a higher prevalence of thrombophilic genotypes in the former group. Notably, the methylene tetrahydrofolate reductase TT677 genotype was present in 50% of patients, the factor V Leiden in 25%, and the prothrombin transition G20210A in 25%, with combined mutations identified in 33% of patients [2]. A retrospective analysis that included

14 original studies and 120 patients showed that the highest pooled percentage of any inherited thrombophilic factor was Factor V Leiden mutation in 9% (CI 2.9 - 16.1) and prothrombin gene mutation in 7% (CI 2.7 - 11.8). The highest pooled percentage of acquired thrombophilia was JAK2 V617F mutation in 14% (CI 1.9 - 28.1) of patients [5].

In our patient, the etiology is multifactorial, with an underlying Factor V Leiden mutation, recent concurrent use of OCPs containing ethinylnorgestrel-levonorgestrel, a 7.5-pack years of smoking history, and morbid obesity. The presentation of MVT is either acute with sudden onset of symptoms or subacute with presentation over days to weeks. Approximately 75% of patients exhibit symptoms for more than 48 hours before seeking medical attention, with reported mean durations ranging from 6 to 14 days [3]. The most common symptom of acute MVT is abdominal pain, which is present in 91% - 100% of cases, additional symptoms may include abdominal distension, nausea, anorexia, vomiting, constipation, and gastrointestinal bleeding, with occult blood detected in 50% of cases [2] [6].

The diagnostic modality of choice is contrast-enhanced CT, with a sensitivity and specificity of 96% and 90% - 94%, respectively [3]-[5] CT imaging also assesses the extent of thrombosis and identifies local precipitating factors; it also identifies complications like bowel gangrene or perforation, which necessitates prompt surgical intervention. CT findings indicative of mesenteric vein thrombosis include visualization of thrombus within the vein, vein expansion with a well-defined wall, and signs of intestinal ischemia such as bowel wall thickening, mesenteric thickening, indistinct bowel margins, and ascites. Homogenous enhancement of the bowel and bowel wall thickening signifies a transmural infarction with a diagnostic accuracy of 90% [3]. One study including 35 MVT patients showed that CT findings predominantly included transmural bowel necrosis (74%), bowel wall thickening (60%), pneumatosis (14%), free fluid (15%), and mesenteric streaking (9%) [6]. Once intestinal infarction has occurred, thumbprinting or pneumatosis may appear; however, mortality is already more than 75% at this point. Routine laboratory tests typically lack diagnostic utility as well. However, severe disease with dehydration may present with hemoconcentration and leukocytosis, while late-stage complications may present with hypoxemia and lactic acidosis, both of which predict a poor prognosis [3]-[7].

Lab testing for thrombophilia, includes screening for factor V Leiden mutation, protein C deficiency, protein S deficiency, antithrombin deficiency, prothrombin G20210A gene mutation, lupus anticoagulant, cardiolipin antibodies, hyperhomocysteinemia, beta-2 glycoprotein antibodies, antiphospholipid syndrome, and JAK2 V617F mutations etc [5]. The main goals of treatment are to prevent thrombus extension in the short term and to prevent the recurrence of thrombosis in the long term. Thus, anticoagulation is the initial approach to management, and Heparin or direct oral anticoagulants (DOACs) should be administered as soon as the diagnosis is made [3].

Anticoagulation has significantly increased survival rates, with studies indicat-

ing recanalization in most patients. Following the acute phase and in the absence of immediate surgical intervention, anticoagulation should be maintained to prevent the recurrence of thrombosis. A recommended duration of 3 - 6 months is advised for patients with reversible causes, while for patients with persistent hypercoagulable state, irreversible systemic condition, or idiopathic cases, lifelong anticoagulation must be considered. Warfarin can maintain anticoagulation with a target INR of 2 - 3. However, frequent monitoring is required.

Alternatively, fixed doses of direct thrombin and factor Xa inhibitors can be used, which require less frequent monitoring and less risk of interactions with other medications. With prolonged anticoagulation, the risk of bleeding must be considered; however, studies have found the risk to be as low as 10% [2]. Some series indicate that a delay in starting 6 - 12 hr anticoagulation increases mortality to 50% - 60% and 80% - 100% if the delay is more than 24 hours [1]-[3].

4. Conclusions

It is imperative to maintain a high index of suspicion when managing patients with acute or subacute onset of abdominal pain that have risk factors for thrombosis. Imaging with contrast-enhanced CT must be obtained to identify thrombosis with prompt initiation of anticoagulant therapy, which should be continued indefinitely in those with underlying and irreversible prothrombotic states to prevent a recurrence. Patients must be monitored closely for response to medical therapy, and timely surgical intervention must be considered in case of persistence of symptoms, or imaging findings of gangrene.

5. Disclosures

Human subjects: Consent was obtained or waived by all participants in this study. Conflicts of interest: All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work.

Other relationships: All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

Conflicts of Interest

The authors declare no conflicts of interest.

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